

CLAIMS

What is claimed is:

- 5 1. A method of identifying individuals having a polymorphism, comprising:
 - a. providing nucleic acid from a subject; and
 - b. detecting the presence of at least one polymorphism in said nucleic
10 acid, said at least one polymorphism selected from the group
consisting of polymorphisms found in SEQ ID Nos:1-3360 and 3361-
7669.
2. The method of Claim 1, further comprising step c) providing a prognosis
to said subject based on the presence or absence of said at least one polymorphism.
- 15 3. The method of Claim 2, wherein said prognosis comprises a genotype
relative risk.
4. The method of Claim 2, wherein said prognosis comprises a population
attributable risk.
- 20 5. The method of Claim 1, wherein said detecting step comprises use of a
hybridization assay.
6. The method of Claim 1, wherein said detecting step comprises use of a
25 TAQMAN assay.
7. The method of Claim 1, wherein said detecting step comprises use of an
invasive cleavage assay.

8. The method of Claim 1, wherein said detecting step comprises use of mass spectroscopy.

5 9. The method of Claim 1, wherein said detecting step comprises use of a microarray.

10. The method of Claim 1, wherein said detecting step comprises use of a polymerase chain reaction.

10 11. The method of Claim 1, wherein said detecting step comprises use of a rolling circle extension assay.

12. The method of Claim 1, wherein said detecting step comprises use of a sequencing assay.

15 13. The method of Claim 1, wherein said detecting step comprises use of a hybridization assay employing a probe complementary to a polymorphism.

20 14. The method of Claim 1, wherein said detecting step comprises use of a bead array assay.

15. The method of Claim 1, wherein said detecting step comprises use of a primer extension assay.

25 16. The method of Claim 1, wherein said detecting step comprises use of an enzyme mismatch cleavage assay.

30 17. The method of Claim 1, wherein said detecting step comprises use of a branched hybridization assay.

18. The method of Claim 1, wherein said detecting step comprises use of a NASBA assay.

19. The method of Claim 1, wherein said detecting step comprises use of a
5 molecular beacon assay.

20. The method of Claim 1, wherein said detecting step comprises use of a cycling probe assay.

10 21. The method of Claim 1, wherein said detecting step comprises use of a ligase chain reaction assay.

22. The method of Claim 1, wherein said detection step comprises use of a sandwich hybridization assay.

15 23. A composition comprising a nucleic acid, said nucleic acid comprising a sequence selected from the group consisting of SEQ ID NO:1-3360 and 3361-7669 or complements thereof.

20 24. The composition of Claim 23, wherein said nucleic acid is 200 or less nucleotides in length.

25 25. The composition of Claim 23, wherein said nucleic acid molecule comprises a label.

26. The composition of Claim 23, wherein said nucleic acid comprises a gene sequence.

27. The composition of Claim 23, wherein said nucleic acid is attached to a
30 solid support.

28. The vector comprising the nucleic acid of Claim 23.

29. A host cell comprising the vector of Claim 28.

5 30. A polypeptide encoded by the nucleic acid of Claim 23.

31. A kit for detecting a polymorphism, comprising at least one reagent that specifically detects a polymorphism in a sequence selected from the group consisting of SEQ ID Nos:3360 and 3361- 7669.

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32. The kit of Claim 31, further comprising instructions for determining whether the subject is at increased risk of having a drug metabolism disorder.

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33. The kit of Claim 31, wherein said at least one reagent comprises a nucleic acid probe.

34. The kit of Claim 31, wherein said kit comprises an in vitro diagnostic detection assay.

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35. The kit of Claim 31, wherein said kit comprises an analyte specific reagent detection assay.

36. The kit of Claim 31, wherein said kit comprises a research-use-only detection assay.

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37. A method for screening subjects for genetic markers associated with drug metabolizing enzyme(s), comprising:

- a) providing a biological sample comprising a nucleic acid from a subject;

- b) testing said nucleic acid for a polymorphism in a genetic marker associated with a drug metabolizing enzyme, said genetic marker comprising one or more nucleotide polymorphisms designated by n, said n selected from a base substitution, an insertion, or a deletion found in a sequence selected from the group consisting of SEQ ID Nos:1-3360 and 3361-7669.

38. The method of Claim 37, wherein said biological sample is selected from the group consisting of blood, saliva, amniotic fluid, and tissue.

39. The method of Claim 37, wherein said subject is a human.

40. The method of Claim 37, wherein said nucleic acid comprises DNA.

41. The method of Claim 37, wherein said nucleic acid comprises RNA.

42. A composition comprising an array of detection assays, said array comprising a plurality of drug metabolizing enzyme nucleotide polymorphism detection assays, one or more of said detection assays being capable of detecting one or more nucleotide polymorphisms designated by n in SEQ ID Nos:1-3360 and 3361-7669, wherein n represents a base substitution, insertion, or deletion compared to a wild-type sequence.

43. The composition of Claim 42, wherein said detection assay is selected from the group consisting of a sequencing assay, a polymerase chain reaction assay, a hybridization assay, a hybridization assay employing a probe complementary to a polymorphism, a microarray assay, a bead array assay, a primer extension assay, an enzyme mismatch cleavage assay, a branched hybridization assay, a rolling circle replication assay, a NASBA assay, a molecular beacon assay, a cycling probe assay, a ligase chain reaction assay, and a sandwich hybridization assay.

44. A composition comprising a detection probe for determining the presense or absence a single nucleotide polymorphism in a gene encoding a drug metabolizing enzyme, said gene comprising a sequence selected from the group consisting of SEQ ID Nos:1-3360 and 3361-7669.

45. A kit comprising the detection probe of Claim 44, and at least one PCR primer for amplifying at least a portion of said gene.

46. A method of determining the effectiveness of or side-effect of a drug or treatment protocol, comprising;

- a. administering a drug or treatment protocol to one or more subjects;
- b. obtaining nucleic acid from said one or more subjects;
- c. using a detection assay to detect the presence of at least one polymorphism in said nucleic acid from said one or more of subjects, said at least one polymorphism selected from the group consisting of polymorphisms found in SEQ ID Nos:1-3360 and 3361-7669; and,
- d. assigning an effectiveness rating, side-effect rating, or score for said drug or treatment protocol based upon a result of one or more said detection assays.

47. The method of Claim 46, wherein said detection assay comprises a hybridization assay.

48. The method of Claim 46, wherein said detection assay comprises a TAQMAN assay.

49. The method of Claim 46, wherein said detection assay comprises an invasive cleavage assay.

50. The method of Claim 46, wherein said detection assay comprises mass spectroscopy.

5 51. The method of Claim 46, wherein said detection assay comprises a microarray.

52. The method of Claim 46, wherein said detection assay comprises a polymerase chain reaction.

10 53. The method of Claim 46, wherein said detection assay comprises a rolling circle extension assay.

15 54. The method of Claim 46, wherein said detection assay comprises a sequencing assay.

55. The method of Claim 46, wherein said detection assay comprises a hybridization assay employing a probe complementary to a polymorphism.

20 56. The method of Claim 46, wherein said detection assay comprises a bead array assay.

57. The method of Claim 46, wherein said detection assay comprises a primer extension assay.

25 58. The method of Claim 46, wherein said detection assay comprises an enzyme mismatch cleavage assay.

30 59. The method of Claim 46, wherein said detection assay comprises a branched hybridization assay.

60. The method of Claim 46, wherein said detection assay comprises a NASBA assay.

5 61. The method of Claim 46, wherein said detection assay comprises a molecular beacon assay.

62. The method of Claim 46, wherein said detection assay comprises a cycling probe assay.

10 63. The method of Claim 46, wherein said detection assay comprises a ligase chain reaction assay.

15 64. The method of Claim 46, wherein said detection step comprises a sandwich hybridization assay.

65. The method of Claim 46, in which said detection assay comprises a kit for detecting a polymorphism, said kit comprising at least one reagent that specifically detects a polymorphism in a sequence selected from the group consisting of SEQ ID
20 Nos:1-3360 and 3361-7669.

66. The method of Claim 65, further comprising instructions for determining whether the subject is at increased risk of having a drug metabolism disorder.

25 67. The method of Claim 65, wherein said at least one reagent comprises a nucleic acid probe.

68. The method of Claim 65, wherein said kit comprises an analyte specific reagent detection assay.

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69. The method of Claim 65, wherein said kit comprises a research-use-only detection assay.

70. The method of Claim 46, wherein said nucleic acid is obtained from a biological sample, said sample being selected from the group consisting of blood, saliva, amniotic fluid, and tissue.

71. The method of Claim 70, wherein said subject is a mammal.

72. The method of Claim 70, wherein said nucleic acid comprises DNA.

73. The method of Claim 70, wherein said nucleic acid comprises RNA.

74. The method of Claim 65, wherein said kit comprises PCR primers.

75. The method of Claim 74, in which said kit comprises an array of detection assays, said array comprising a plurality of drug metabolizing enzyme nucleotide polymorphism detection assays, one or more of said detection assays being capable of detecting one or more nucleotide polymorphisms designated by n in SEQ ID Nos:1-3360 and 3361-7669, wherein n represents a base substitution, insertion, or deletion compared to a wild-type sequence.

76. A method of prescribing a drug to or treatment protocol for a subject, comprising;

- a. providing nucleic acid from said subject;
- b. using a detection assay to detect the presence of at least one polymorphism in said nucleic acid, said at least one polymorphism selected from the group consisting of polymorphisms found in SEQ ID Nos:1-3360 and 3361-7669; and,

c. prescribing said drug or treatment protocol based upon the result of said detection assay.

77. The method of Claim 76, further comprising step d) providing a prognosis to said subject based on the presence or absence of said at least one polymorphism.

78. The method of Claim 77, wherein said prognosis comprises a genotype relative risk.

79. The method of Claim 77, wherein said prognosis comprises a population attributable risk.

80. The method of Claim 76, wherein said detection assay comprises a hybridization assay.

81. The method of Claim 76, wherein said detection assay comprises a TAQMAN assay.

82. The method of Claim 76, wherein said detection assay comprises an invasive cleavage assay.

83. The method of Claim 76, wherein said detection assay comprises mass spectroscopy.

84. The method of Claim 76, wherein said detection assay comprises a microarray.

85. The method of Claim 76, wherein said detection assay comprises a polymerase chain reaction.

86. The method of Claim 76, wherein said detection assay comprises a rolling circle extension assay.

87. The method of Claim 76, wherein said detection assay comprises a
5 sequencing assay.

88. The method of Claim 76, wherein said detection assay comprises a hybridization assay employing a probe complementary to a polymorphism.

10 89. The method of Claim 76, wherein said detection assay comprises a bead array assay.

90. The method of Claim 76, wherein said detection assay comprises a primer extension assay.

15 91. The method of Claim 76, wherein said detection assay comprises an enzyme mismatch cleavage assay.

20 92. The method of Claim 76, wherein said detection assay comprises a branched hybridization assay.

93. The method of Claim 76, wherein said detection assay comprises a NASBA assay.

25 94. The method of Claim 76, wherein said detection assay comprises a molecular beacon assay.

95. The method of Claim 76, wherein said detection assay comprises a cycling probe assay.

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96. The method of Claim 76, wherein said detection assay comprises a ligase chain reaction assay.

97. The method of Claim 76, wherein said detection step comprises a sandwich hybridization assay.

98. The method of Claim 76, in which said detection assay comprises a kit for detecting a polymorphism, said kit comprising at least one reagent that specifically detects a polymorphism in a sequence selected from the group consisting of SEQ ID Nos:1-3360 and 3361-7669.

99. The method of Claim 98, further comprising instructions for determining whether the subject is at increased risk of having a drug metabolism disorder.

100. The method of Claim 99, wherein said at least one reagent comprises a nucleic acid probe.

101. The method of Claim 98, wherein said kit comprises an in vitro diagnostic detection assay.

102. The method of Claim 98, wherein said kit comprises an analyte specific reagent detection assay.

103. The method of Claim 98, wherein said kit comprises a research-use-only detection assay.

104. The method of Claim 76, wherein said nucleic acid is obtained from a biological sample, said sample being selected from the group consisting of blood, saliva, amniotic fluid, and tissue.

105. The method of Claim 104, wherein said subject is a human.

106. The method of Claim 104, wherein said nucleic acid comprises DNA.

5 107. The method of Claim 104, wherein said nucleic acid comprises RNA.

108. The method of Claim 104, wherein said kit comprises PCR primers.

109. The method of claim 98, in which said kit comprises an array of detection
10 assays, said array comprising a plurality of drug metabolizing enzyme nucleotide
polymorphism detection assays, one or more of said detection assays being capable of
detecting one or more nucleotide polymorphisms designated by n in SEQ ID Nos:1-3360
and 3361-7669, wherein n represents a base substitution, insertion, or deletion compared
to a wild-type sequence.

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110. The method of Claim 109, wherein said detection assay is selected from
the group consisting of a sequencing assay, a polymerase chain reaction assay, a
hybridization assay, a hybridization assay employing a probe complementary to a
polymorphism, a microarray assay, a bead array assay, a primer extension assay, an
20 enzyme mismatch cleavage assay, a branched hybridization assay, a rolling circle
replication assay, a NASBA assay, a molecular beacon assay, a cycling probe assay, a
ligase chain reaction assay, and a sandwich hybridization assay.

111. A method for generating assay data comprising:
25 a. obtaining a sample from a subject containing nucleic acid;
b. transferring said sample to a laboratory; and
c. receiving data from said laboratory, wherein said data corresponds to
the presence of at least one polymorphism in said nucleic acid, said at
least one polymorphism selected from the group consisting of
30 polymorphisms found in SEQ ID Nos:1-3360 and 3361-7669.

112. A data set generated by the method of Claim 111.

113. A composition comprising a nucleic acid, said nucleic acid comprising a
5 gene sequence, said gene sequence having at least one polymorphism, said at least one
polymorphism selected from the group consisting of polymorphisms found in SEQ ID
NOs:1-3360 and 3361-7669.

114. The composition of Claim 113, wherein said at least one polymorphism is
10 located in a non-coding portion of said gene.

115. The composition of Claim 113, wherein said at least one polymorphism is
located in an intron.

116. The composition of Claim 113, wherein said at least one polymorphism is
15 located in an exon.

117. The composition of Claim 113, wherein said at least one polymorphism is
located in a 5' untranslated portion of said gene.
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118. The composition of Claim 113, wherein said at least one polymorphism is
located in a 3' untranslated portion of said gene.

119. The composition of Claim 113, wherein said at least one polymorphism is
25 located in an untranslated regulatory portion of said gene.

120. The composition of Claim 113, wherein said at least one polymorphism is
located in a region of said gene affecting splicing.

121. The composition of Claim 113, wherein said at least one polymorphism is located in a region of said gene affecting level of transcription of said gene.

122. The composition of Claim 113, wherein two or more of said
5 polymorphisms are located in a gene sequence, said gene sequence comprising an intron, an exon, a 5' untranslated portion of said gene, a 3' untranslated portion of said gene, a regulatory portion of said gene, a portion of said gene affecting splicing, and a portion of said gene affecting level or transcription of said gene.

10 123. A composition comprising an oligonucleotide, said oligonucleotide comprising at least one polymorphism, said at least one polymorphism selected from the group consisting of polymorphisms found in SEQ ID Nos:1-3360 and 3361-7669, and said oligonucleotide comprising at least five bases upstream or five bases downstream of said polymorphism.

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